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RAW SEQUENCE LISTING
PATENT APPLICATION: US/09/735,995

DATE: 04/03/2001
TIME: 12:35:42

2.

Input Set : N:\Crf3\RULE60\09735995.txt
Output Set: N:\CRF3\04032001\I735995.raw

5 <110> APPLICANT: Keating, Mark T.
7 Splawski, Igor
11 <120> TITLE OF INVENTION: MUTATIONS IN AND GENOMIC STRUCTURE OF HERG - A LONG QT
13 SYNDROME GENE
17 <130> FILE REFERENCE: 2323-136
21 <140> CURRENT APPLICATION NUMBER: 09/735,995
23 <141> CURRENT FILING DATE: 2000-12-14
27 <150> PRIOR APPLICATION NUMBER: 09/226,012
28 <151> PRIOR FILING DATE: 1999-01-06
31 <160> NUMBER OF SEQ ID NOS: 116
35 <170> SOFTWARE: PatentIn Ver. 2.0
39 <210> SEQ ID NO: 1
41 <211> LENGTH: 3480
43 <212> TYPE: DNA
45 <213> ORGANISM: Homo sapiens
49 <220> FEATURE:
51 <221> NAME/KEY: CDS
53 <222> LOCATION: (1)..(3477)
57 <400> SEQUENCE: 1

ENTERED

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63 1 5 10 15
67 acc atc atc cgc aag ttt gag ggc cag agc cgt aag ttc atc atc gcc 96
69 Thr Ile Ile Arg Lys Phe Glu Gly Gln Ser Arg Lys Phe Ile Ile Ala
71 20 25 30
75 aac gct cgg gtg gag aac tgc gcc gtc atc tac tgc aac gac ggc ttc 144
77 Asn Ala Arg Val Glu Asn Cys Ala Val Ile Tyr Cys Asn Asp Gly Phe
79 35 40 45
83 tgc gag ctg tgc ggc tac tgc cgg gcc gag gtg atg cag cga ccc tgc 192
85 Cys Glu Leu Cys Gly Tyr Ser Arg Ala Glu Val Met Gln Arg Pro Cys
87 50 55 60
91 acc tgc gac ttc ctg cac ggg ccg cgc acg cag cgc gct gcc gcg 240
93 Thr Cys Asp Phe Leu His Gly Pro Arg Thr Gln Arg Arg Ala Ala Ala
95 65 70 75 80
99 cag atc gcg cag gca ctg ctg ggc gcc gag gag cgc aaa gtg gaa atc 288
101 Gln Ile Ala Gln Ala Leu Leu Gly Ala Glu Glu Arg Lys Val Glu Ile
103 85 90 95
107 gcc ttc tac cgg aaa gat ggg agc tgc ttc cta tgt ctg gtg gat gtg 336
109 Ala Phe Tyr Arg Lys Asp Gly Ser Cys Phe Leu Cys Leu Val Asp Val
111 100 105 110
115 gtg ccc gtg aag aac gag gat ggg gct gtc atc atg ttc atc ctc aat 384
117 Val Pro Val Lys Asn Glu Asp Gly Ala Val Ile Met Phe Ile Leu Asn
119 115 120 125
123 ttc gag gtg gtg atg gag aag gac atg gtg ggg tcc ccg gct cat gac 432
125 Phe Glu Val Val Met Glu Lys Asp Met Val Gly Ser Pro Ala His Asp
127 130 135 140
131 acc aac cac cgg ggc ccc ccc acc agc tgg ctg gcc cca ggc cgc gcc 480

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133 Thr Asn His Arg Gly Pro Pro Thr Ser Trp Leu Ala Pro Gly Arg Ala
135 145 150 155 160
139 aag acc ttc cgc ctg aag ctg ccc gcg ctg ctg gcg ctg acg gcc cgg 528
141 Lys Thr Phe Arg Leu Lys Leu Pro Ala Leu Leu Ala Leu Thr Ala Arg
143 165 170 175
147 gag tcg tcg gtg cgg tcg ggc ggc gcg ggc ggc gcg gcc ccg ggg 576
149 Glu Ser Ser Val Arg Ser Gly Gly Ala Gly Gly Ala Gly Ala Pro Gly
151 180 185 190
155 gcc gtg gtg gtg gac gtg gac ctg acg ccc gcg gca ccc agc agc gag 624
157 Ala Val Val Val Asp Val Asp Leu Thr Pro Ala Ala Pro Ser Ser Glu
159 195 200 205
163 tcg ctg gcc ctg gac gaa gtg aca gcc atg gac aac cac gtg gca ggg 672
165 Ser Leu Ala Leu Asp Glu Val Thr Ala Met Asp Asn His Val Ala Gly
167 210 215 220
171 ctc ggg ccc gcg gag gag cgg cgt gcg ctg gtg ggt ccc ggc tct ccg 720
173 Leu Gly Pro Ala Glu Glu Arg Arg Ala Leu Val Gly Pro Gly Ser Pro
175 225 230 235 240
179 ccc cgc agc gcg ccc ggc cag ctc cca tcg ccc cgg gcg cac agc ctc 768
181 Pro Arg Ser Ala Pro Gly Gln Leu Pro Ser Pro Arg Ala His Ser Leu
183 245 250 255
187 aac ccc gac gcc tcg ggc tcc agc tgc agc ctg gcc cgg acg cgc tcc 816
189 Asn Pro Asp Ala Ser Gly Ser Ser Cys Ser Leu Ala Arg Thr Arg Ser
191 260 265 270
195 cga gaa agc tgc gcc agc gtg cgc gcg gcc tcg tcg gcc gac gac atc 864
197 Arg Glu Ser Cys Ala Ser Val Arg Arg Ala Ser Ser Ala Asp Asp Ile
199 275 280 285
203 gag gcc atg cgc gcc ggg gtg ctg ccc ccg cca ccg cgc cac gcc agc 912
205 Glu Ala Met Arg Ala Gly Val Leu Pro Pro Pro Pro Arg His Ala Ser
207 290 295 300
211 acc ggg gcc atg cac cca ctg cgc agc ggc ttg ctc aac tcc acc tcg 960
213 Thr Gly Ala Met His Pro Leu Arg Ser Gly Leu Leu Asn Ser Thr Ser
215 305 310 315 320
219 gac tcc gac ctc gtg cgc tac cgc acc att agc aag att ccc caa atc 1008
221 Asp Ser Asp Leu Val Arg Tyr Arg Thr Ile Ser Lys Ile Pro Gln Ile
223 325 330 335
227 acc ctc aac ttt gtg gac ctc aag ggc gac ccc ttc ttg gct tcg ccc 1056
229 Thr Leu Asn Phe Val Asp Leu Lys Gly Asp Pro Phe Leu Ala Ser Pro
231 340 345 350
235 acc agt gac cgt gag atc ata gca cct aag ata aag gag cga acc cac 1104
237 Thr Ser Asp Arg Glu Ile Ile Ala Pro Lys Ile Lys Glu Arg Thr His
239 355 360 365
243 aat gtc act gag aag gtc acc cag gtc ctg tcc ctg ggc gcc gac gtg 1152
245 Asn Val Thr Glu Lys Val Thr Gln Val Leu Ser Leu Gly Ala Asp Val
247 370 375 380
251 ctg cct gag tac aag ctg cag gca ccg cgc atc cac cgc tgg acc atc 1200
253 Leu Pro Glu Tyr Lys Leu Gln Ala Pro Arg Ile His Arg Trp Thr Ile
255 385 390 395 400
259 ctg cat tac agc ccc ttc aag gcc gtg tgg gac tgg ctc atc ctg ctg 1248
261 Leu His Tyr Ser Pro Phe Lys Ala Val Trp Asp Trp Leu Ile Leu Leu

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263	405	410	415	1296
267	ctg gtc atc tac acg gct gtc ttc aca ccc tac tcg gct gcc ttc ctg			
269	Leu Val Ile Tyr Thr Ala Val Phe Thr Pro Tyr Ser Ala Ala Phe Leu			
271	420	425	430	
275	ctg aag gag acg gaa gaa ggc ccg cct gct acc gag tgt gcc tac gcc			1344
277	Leu Lys Glu Thr Glu Glu Gly Pro Pro Ala Thr Glu Cys Gly Tyr Ala			
279	435	440	445	
283	tgc cag ccg ctg gct gtg gtg gac ctc atc gtg gac atc atg ttc att			1392
285	Cys Gln Pro Leu Ala Val Val Asp Leu Ile Val Asp Ile Met Phe Ile			
287	450	455	460	
291	gtg gac atc ctc atc aac ttc cgc acc acc tac gtc aat gcc aac gag			1440
293	Val Asp Ile Leu Ile Asn Phe Arg Thr Thr Tyr Val Asn Ala Asn Glu			
295	465	470	475	480
299	gag gtg gtc agc cac ccc ggc cgc atc gcc gtc cac tac ttc aag ggc			1488
301	Glu Val Val Ser His Pro Gly Arg Ile Ala Val His Tyr Phe Lys Gly			
303	485	490	495	
307	tgg ttc ctc atc gac atg gtg gcc gcc atc ccc ttc gac ctg ctc atc			1536
309	Trp Phe Leu Ile Asp Met Val Ala Ala Ile Pro Phe Asp Leu Leu Ile			
311	500	505	510	
315	ttc ggc tct ggc tct gag gag ctg atc ggg ctg ctg aag act gcg cgg			1584
317	Phe Gly Ser Gly Ser Glu Glu Ile Gly Leu Leu Lys Thr Ala Arg			
319	515	520	525	
323	ctg ctg cgg ctg gtg cgc gtg gcg cgg aag ctg gat cgc tac tca gag			1632
325	Leu Leu Arg Leu Val Arg Val Ala Arg Lys Leu Asp Arg Tyr Ser Glu			
327	530	535	540	
331	tac ggc gcg gcc gtg ctg ttc ttg ctc atg tgc acc ttt gcg ctc atc			1680
333	Tyr Gly Ala Ala Val Leu Phe Leu Leu Met Cys Thr Phe Ala Leu Ile			
335	545	550	555	560
339	gcg cac tgg cta gcc tgc atc tgg tac gcc atc ggc aac atg gag cag			1728
341	Ala His Trp Leu Ala Cys Ile Trp Tyr Ala Ile Gly Asn Met Glu Gln			
343	565	570	575	
347	cca cac atg gac tca cgc atc ggc tgg ctg cac aac ctg ggc gac cag			1776
349	Pro His Met Asp Ser Arg Ile Gly Trp Leu His Asn Leu Gly Asp Gln			
351	580	585	590	
355	ata ggc aaa ccc tac aac agc agc ggc ctg ggc ggc ccc tcc atc aag			1824
357	Ile Gly Lys Pro Tyr Asn Ser Ser Gly Leu Gly Gly Pro Ser Ile Lys			
359	595	600	605	
363	gac aag tat gtg acg gcg ctc tac ttc acc ttc agc agc ctc acc agt			1872
365	Asp Lys Tyr Val Thr Ala Leu Tyr Phe Thr Phe Ser Ser Leu Thr Ser			
367	610	615	620	
371	gtg ggc ttc ggc aac gtc tct ccc aac acc aac tca gag aag atc ttc			1920
373	Val Gly Phe Gly Asn Val Ser Pro Asn Thr Asn Ser Glu Lys Ile Phe			
375	625	630	635	640
379	tcc atc tgc gtc atg ctc att ggc tcc ctc atg tat gct agc atc ttc			1968
381	Ser Ile Cys Val Met Leu Ile Gly Ser Leu Met Tyr Ala Ser Ile Phe			
383	645	650	655	
387	ggc aac gtg tcg gcc atc atc cag cgg ctg tac tcg ggc aca gcc cgc			2016
389	Gly Asn Val Ser Ala Ile Ile Gln Arg Leu Tyr Ser Gly Thr Ala Arg			
391	660	665	670	

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395	tac	cac	aca	cag	atg	ctg	cgg	gtg	cgg	gag	ttc	atc	cgc	ttc	cac	cag	2064
397	Tyr	His	Thr	Gln	Met	Leu	Arg	Val	Arg	Glu	Phe	Ile	Arg	Phe	His	Gln	
399			675					680					685				
403	atc	ccc	aat	ccc	ctg	cgc	cag	cgc	ctc	gag	gag	tac	ttc	cag	cac	gcc	2112
405	Ile	Pro	Asn	Pro	Leu	Arg	Gln	Arg	Leu	Glu	Glu	Tyr	Phe	Gln	His	Ala	
407		690					695						700				
411	tgg	tcc	tac	acc	aac	ggc	atc	gac	atg	aac	gcg	gtg	ctg	aag	ggc	ttc	2160
413	Trp	Ser	Tyr	Thr	Asn	Gly	Ile	Asp	Met	Asn	Ala	Val	Leu	Lys	Gly	Phe	
415	705					710				715							
419	cct	gag	tgc	ctg	cag	gct	gac	atc	tgc	ctg	cac	ctg	aac	cgc	tca	ctg	2208
421	Pro	Glu	Cys	Leu	Gln	Ala	Asp	Ile	Cys	Leu	His	Leu	Asn	Arg	Ser	Leu	
423						725				730				735			
427	ctg	cag	cac	tgc	aaa	ccc	ttc	cga	ggg	gcc	acc	aag	ggc	tgc	ctt	cgg	2256
429	Leu	Gln	His	Cys	Lys	Pro	Phe	Arg	Gly	Ala	Thr	Lys	Gly	Cys	Leu	Arg	
431						740				745				750			
435	gcc	ctg	gcc	atg	aag	ttc	aag	acc	aca	cat	gca	ccg	cca	ggg	gac	aca	2304
437	Ala	Leu	Ala	Met	Lys	Phe	Lys	Thr	His	Ala	Pro	Pro	Gly	Asp	Thr		
439						755				760				765			
443	ctg	gtg	cat	gct	ggg	gac	ctg	ctc	acc	gcc	ctg	tac	ttc	atc	tcc	cgg	2352
445	Leu	Val	His	Ala	Gly	Asp	Leu	Leu	Thr	Ala	Leu	Tyr	Phe	Ile	Ser	Arg	
447		770				775						780					
451	ggc	tcc	atc	gag	atc	ctg	cgg	ggc	gac	gtc	gtc	gtg	gcc	atc	ctg	ggg	2400
453	Gly	Ser	Ile	Glu	Ile	Leu	Arg	Gly	Asp	Val	Val	Val	Ala	Ile	Leu	Gly	
455	785					790				795						800	
459	aag	aat	gac	atc	ttt	ggg	gag	cct	ctg	aac	ctg	tat	gca	agg	cct	ggc	2448
461	Lys	Asn	Asp	Ile	Phe	Gly	Glu	Pro	Leu	Asn	Leu	Tyr	Ala	Arg	Pro	Gly	
463						805				810						815	
467	aag	tcg	aac	ggg	gat	gtg	cgg	gcc	ctc	acc	tac	tgt	gac	cta	cac	aag	2496
469	Lys	Ser	Asn	Gly	Asp	Val	Arg	Ala	Leu	Thr	Tyr	Cys	Asp	Leu	His	Lys	
471						820				825				830			
475	atc	cat	cgg	gac	gac	ctg	ctg	gag	gtg	ctg	gac	atg	tac	cct	gag	ttc	2544
477	Ile	His	Arg	Asp	Asp	Leu	Leu	Glu	Val	Leu	Asp	Met	Tyr	Pro	Glu	Phe	
479						835				840				845			
483	tcc	gac	cac	ttc	tgg	tcc	agc	ctg	gag	atc	acc	ttc	aac	ctg	cga	gat	2592
485	Ser	Asp	His	Phe	Trp	Ser	Ser	Leu	Glu	Ile	Thr	Phe	Asn	Leu	Arg	Asp	
487						850				855				860			
491	acc	aac	atg	atc	ccg	ggc	tcc	ccc	ggc	agt	acg	gag	tta	gag	ggt	ggc	2640
493	Thr	Asn	Met	Ile	Pro	Gly	Ser	Pro	Gly	Ser	Thr	Glu	Leu	Glu	Gly	Gly	
495	865					870				875						880	
499	ttc	agt	cgg	caa	cgc	aag	cgc	aag	ttg	tcc	ttc	cgc	agg	cgc	acg	gac	2688
501	Phe	Ser	Arg	Gln	Arg	Lys	Arg	Lys	Leu	Ser	Phe	Arg	Arg	Arg	Thr	Asp	
503						885				890						895	
507	aag	gac	acg	gag	cag	cca	ggg	gag	gtg	tcg	gcc	ttg	ggg	ccg	ggc	cgg	2736
509	Lys	Asp	Thr	Glu	Gln	Pro	Gly	Glu	Val	Ser	Ala	Leu	Gly	Pro	Gly	Arg	
511						900				905				910			
515	gcg	ggg	gca	ggg	ccg	agt	agc	cgg	ggc	cgg	ccg	ggg	ggg	ccg	tgg	ggg	2784
517	Ala	Gly	Ala	Gly	Pro	Ser	Ser	Arg	Gly	Arg	Pro	Gly	Gly	Pro	Trp	Gly	
519						915				920				925			
523	gag	agc	ccg	tcc	agt	ggc	ccc	tcc	agc	cct	gag	agc	agt	gag	gat	gag	2832

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525 Glu Ser Pro Ser Ser Gly Pro Ser Ser Pro Glu Ser Ser Glu Asp Glu
527      930      935      940
531 ggc cca ggc cgc agc tcc agc ccc ctc cgc ctg gtg ccc ttc tcc agc 2880
533 Gly Pro Gly Arg Ser Ser Ser Pro Leu Arg Leu Val Pro Phe Ser Ser
535 945      950      955      960
539 ccc agg ccc ccc gga gag ccg ccg ggt ggg gag ccc ctg atg gag gac 2928
541 Pro Arg Pro Pro Gly Glu Pro Pro Gly Gly Glu Pro Leu Met Glu Asp
543      965      970      975
547 tgc gag aag agc agc gac act tgc aac ccc ctg tca ggc gcc ttc tca 2976
549 Cys Glu Lys Ser Ser Asp Thr Cys Asn Pro Leu Ser Gly Ala Phe Ser
551      980      985      990
555 gga gtg tcc aac att ttc agc ttc tgg ggg gac agt cgg ggc cgc cag 3024
557 Gly Val Ser Asn Ile Phe Ser Phe Trp Gly Asp Ser Arg Gly Arg Gln
559      995      1000      1005
563 tac cag gag ctc cct cga tgc ccc gcc ccc acc ccc agc ctc ctc aac 3072
565 Tyr Gln Glu Leu Pro Arg Cys Pro Ala Pro Thr Pro Ser Leu Leu Asn
567      1010      1015      1020
571 atc ccc ctc tcc agc ccg ggt ccg ccg ccc ccg ggc gac gtg gag agc 3120
573 Ile Pro Leu Ser Ser Pro Gly Arg Arg Pro Arg Gly Asp Val Glu Ser
575 1025      1030      1035
579 agg ctg gat gcc ctc cag cgc cag ctc aac agg ctg gag acc ccg ctg 3168
581 Arg Leu Asp Ala Leu Gln Arg Gln Leu Asn Arg Leu Glu Thr Arg Leu
583      1045      1050      1055
587 agt gca gac atg gcc act gtc ctg cag ctg cta cag agg cag atg acg 3216
589 Ser Ala Asp Met Ala Thr Val Leu Gln Leu Leu Gln Arg Gln Met Thr
591      1060      1065      1070
595 ctg gtc ccg ccc gcc tac agt gct gtg acc acc ccg ggg cct ggc ccc 3264
597 Leu Val Pro Pro Ala Tyr Ser Ala Val Thr Thr Pro Gly Pro Gly Pro
599      1075      1080      1085
603 act tcc aca tcc ccg ctg ttg ccc gtc agc ccc ctc ccc acc ctc acc 3312
605 Thr Ser Thr Ser Pro Leu Leu Pro Val Ser Pro Leu Pro Thr Leu Thr
607      1090      1095      1100
611 ttg gac tcg ctt tct cag gtt tcc cag ttc atg gcg tgt gag gag ctg 3360
613 Leu Asp Ser Leu Ser Gln Val Ser Gln Phe Met Ala Cys Glu Glu Leu
615 1105      1110      1115      1120
619 ccc ccg ggg gcc cca gag ctt ccc caa gaa ggc ccc aca cga cgc ctc 3408
621 Pro Pro Gly Ala Pro Glu Leu Pro Gln Glu Gly Pro Thr Arg Arg Leu
623      1125      1130      1135
627 tcc cta ccg ggc cag ctg ggg gcc ctc acc tcc cag ccc ctg cac aga 3456
629 Ser Leu Pro Gly Gln Leu Gly Ala Leu Thr Ser Gln Pro Leu His Arg
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647 <211> LENGTH: 1159
649 <212> TYPE: PRT
651 <213> ORGANISM: Homo sapiens
655 <400> SEQUENCE: 2

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VERIFICATION SUMMARY
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